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6 Social cohesion

6.2 European Health Committee (CDSP)

c. Recommendation CM/Rec(2010)11 of the Committee of Ministers to member states on the impact of genetics on the organisation of health care services and training of health professionals – Explanatory Memorandum

Recommendation CM/Rec(2010)11 of the Committee of Ministers to member states on the impact of genetics on the organisation of health care services and training of health professionals

EXPLANATORY MEMORANDUM

Introduction

1. Rapid developments in human genetics, especially of molecular genetics, such as the sequencing of the human genome (Human Genome Project) are having a profound impact on health care services. Advances in knowledge of human genetics have led to a better understanding of the molecular basis of disease and has consequently resulted in improved clinical diagnosis and in novel approaches to prevention and treatment of genetic diseases. In parallel, the accompanying wide public awareness of these developments has raised expectations regarding more accurate assessment of genetic risk of individuals and within families and treatment of genetic disease. The challenge lies in ensuring implementation by member states of the developments in genetic diseases so that the benefits will be available to all.

2. Genetic diseases contribute to a significant proportion of health problems. About 5.5% of the population are expected to develop a genetic or partly genetic condition by the age of 25. Many admissions to paediatric hospitals are due to genetic and developmental disorders. The majority of the patients who consult primary care physicians and clinical specialists have diseases that are in part due to genetic factors (multifactorial diseases).

3. Several thousand diseases are known to be associated with genetic mutations. A number of those diseases are associated with a mutation at a single gene locus (monogenic disease) and follow a well-defined pattern of inheritance in the family (Mendelian inheritance). These include diseases such as cystic fibrosis, muscular dystrophies and haemophilia. Although each monogenic condition is in general rare, taken together they affect about 1/100 people. An important category of disease results from environmental trigger factors interacting with a specific genetic predisposition (multifactorial disease). These diseases include many common chronic diseases, such as hypertension, diabetes mellitus, coronary artery disease and most cancers. Their incidence is in the order of 10/100. Chromosomal aberrations are seen in about 1/1000 people. Finally there are other genetic conditions such as mitochondrial disease and parental imprinting disorders of which the incidence is yet unclear.

4. Genetic services in Europe are founded on world leading scientific genetic research. The results of this research are applied in many centres, particularly the use of genetic tests based on molecular technology, which has rapidly increased over recent years. Overall some 30 million people in Europe suffer from a genetic disease. All 47 member states are experiencing the important burden of identified genetic disease, which has been estimated to cost them 500 million Euros.

5. In 1997, a comparative study of genetic services in Europe by the Concerted Action of Genetic Services in Europe (CAGSE), found genetic service practices and facilities of different countries varied considerably.

6. In the same year, the European Society of Human Genetics (ESHG) embarked on a project to develop professional guidelines on various aspects of human genetics. One aspect of the work was the production of guidelines for genetic services in Europe, published in the European Journal of Human Genetics (2003) 11, Suppl.2. This involved the definition and the aims of genetic services, their organisation, their quality and the role of public education.

7. More recently, a European Union funded project (EuroGentest) has been created to address some of the challenges of the genomic era by encouraging the creation of a European Network of Excellence on genetic testing. It is hoped to develop the essential infrastructure, resources, guidelines, procedures and data-bases of tests. The main objective is to harmonise and improve the quality of European genetic services of molecular, cytogenetic and biochemical tests, covering all aspects of genetic testing - quality management, information databases, public health, new technologies and education.

8. Because of the marked diversity of genetic services throughout Europe, governments should renew their commitment to ensuring that there is appropriate access to, quality and equity of service provision. To that end, education and training as well as research are important; regulations and quality control of genetic testing and screening must be addressed and economic appraisal and public awareness must also be discussed.

9. The populations of member states should have equitable and transparent access to genetic services of a high standard, according to national law. The factors limiting the full enjoyment of and access to genetic services are linked to the low number of trained geneticists and counsellors and costly laboratory equipment.

10. The present Recommendation addresses the issues mentioned above. It is based on the Council of Europe's Convention on Human Rights and Biomedicine and its Additional Protocol concerning genetic testing for health purposes. It also takes into account Recommendation Rec(90)13 on prenatal screening, prenatal genetic diagnosis and associated genetic counselling, Recommendation Rec(92)3 on genetic testing and screening for health care purposes and Recommendation Rec(97)5 on the protection of medical data.

11. Member states may use this Recommendation to review best practice in areas beyond those covered by the Additional Protocol concerning genetic testing for health purposes. In particular the Recommendation proposes policy guidelines in the following areas:

- organisation and quality of genetic services;
- education and training of health care professionals;
- promotion of research in the field of medical genetics and genetic services;
- organisation of genetic screening programmes.

The objective of developing a common framework of European guidelines was to promote the appropriate use of genetic services, integrated within the overall health care setting.

The existing experience was taken into account in order to use the collected evidence to propose an integrated approach in the 47 Council of Europe member states.

The implications of genetic testing and follow-up treatment for the organisation, functioning and assessment of health care services were considered, in particular from the perspective of their utility, cost effectiveness and availability.

I. Genetic services

12. The purpose of the genetic services (public or private) is to respond to the needs of individuals and families wishing to know whether they are at risk of developing or transmitting a disease or disorder with a genetic component, or who are faced with such a disease or disorder. Services include in particular:

a) assessment of persons who are affected or are at risk of a genetic disease, to make the clinical diagnosis, to treat (if possible) or to support, to document and analyse the pedigree, and to estimate the risk of transmission in the family and to provide them genetic counselling, including information concerning possible preventive measures;

b) identification of relatives who are at risk for genetic disorders and to provide them with support and genetic counselling including information concerning possible preventive measures;

c) provision of information and support to other clinicians managing all types of genetic disorders.

13. Genetic services include an integrated clinical and laboratory service. It should provide:

a) clinical and laboratory diagnosis using a range of genetic tests (cytogenetic, molecular and biochemical genetic tests);

b) a calculation of the risk of developing a genetic disorder or of transmitting a genetic condition to children;

c) genetic counselling, including information on possible preventive measures;

d) provision of information for patients, family members and other health care professionals;

e) training and education for health care professionals;

f) review of patients and their families as new scientific developments provide new information and further tests become available.

14. Genetic counselling is a communication and support process aiming to enable individuals and, where appropriate, families to make informed choices with regard to a genetic test and its implications. It deals with the occurrences or risk of occurrence, of a genetic disorder in a family. The process involves appropriately trained health care professionals assisting the individual and/or family by:

a) explaining a clinical diagnosis so that the medical facts about the disorder can be easily understood and to consider the disease's future progress and possible approaches to treatment and management;

b) explaining how hereditary factors may play a role in the condition and the possible risk of occurrence in relatives;

c) explaining the various approaches for dealing with the risk, while minimising psychological distress;

d) explaining the best possible adjustment to the presence of the disorder in an affected family member and/or risk of recurrence of the disorder including when it comes to making procreation choices.

e) communicating with the person concerned in an interactive and detailed manner, taking care to adapt the content of the information provided to the ability of the person to comprehend the often complex genetic context.

15. At community level, genetic services should be organised in such a way as to make available according to national law prenatal diagnosis and newborn screening as well as carrier or other genetic screening. Organisation of genetic screening programmes is addressed in Chapter V.

16. Genetic services should provide support to other clinical specialties, as genetic factors have been identified as having an important role in many diseases, including rare disorders, conditions with complex inheritance and in multifactorial disorders. Genetic services should be organised to provide timely diagnostic assistance to other physicians.

17. It is anticipated that pharmacogenomic developments will improve and rationalise drug treatment for several diseases, by testing and screening for the genetic variation of responses to medicaments. It is a developing field and it will become more important in the future.

18. Genetic centres should develop an active interest in specific categories of genetic disorders, so as to have access to reliable clinical and laboratory information about specific conditions. They should be encouraged to cultivate international co-operation for rare disorders.

19. Genetic centres may have a role in public health genetics. Public health genetics applies the results of advances in human genetics, genomics, and molecular biotechnology to improve public health and prevent disease.

20. A genetic service depends on the expertise of a multidisciplinary team in the delivery of clinical and laboratory services for a wide range of genetic disorders. The multidisciplinary team should comprise clinical geneticists, genetic nurses, non-medical doctor genetic counsellors, and other health care professionals such as psychologists and social workers. Member states should endeavour to harmonise as far as possible the training of all these health care professionals.

21. Primary care is a standard term, referring *inter alia* to the activity of a health care provider who acts as a first point of consultation for all patients. Continuity of care is a key characteristic of primary care.

Primary care is an entry point to a health care system that includes secondary care (by community hospitals) and tertiary care (by medical centres and teaching hospitals). Primary care is provided by certain clinicians, for example, general practitioners, family doctors, paediatricians, etc.

Primary care is characterised by first contact, accessibility, longitudinality, and comprehensiveness; it includes health promotion, disease prevention, health maintenance, counselling, patient education, diagnosis and treatment of acute and chronic illnesses in a variety of health care settings (e.g. office, inpatient, critical care, long-term care, home care, day care, etc.). Primary care is performed and managed by a personal physician often collaborating with other health professionals, and utilising consultation or referral as appropriate.

22. Genetic services are not different from other medical activities and their added value should be assessed by standard methods of economic appraisal and health technology assessment, including the effectiveness and the potential benefits.

II. Education and training of health care professionals

23. Appropriate education and training should be provided for all healthcare professionals. From the results of a comparative study of genetic services in Europe it became clear that comparison of genetic education was complicated by the variety of health care systems, organisational structures in health professional education and health professionals involved at first patient contact. One of the main recommendations of the study was the provision of a joint education and training programme to promote teaching and training programmes in medical genetics to medical and other students in related fields with assessment for specialists, medical geneticists and other health care workers. Besides the scientific and medical education, social, legal and ethical aspects of medical genetics should also be taught. Medical genetics should not be regarded as one of the sub-specialities which might not be assimilated by the health care professional but as a core component of training of all health care professionals. In 2001, the European Society of Human Genetics recommended a formal recognition of medical genetics as a medical speciality in Europe.

24. During preclinical education both lectures and practical courses of genetics should be delivered by an instructor who is a specialist in medical genetics. The basic training in genetics should be directed towards the application of genetics in medicine. The clinical education should include lectures and training in small groups of students that allow for interaction between the teacher and the student.

25. The essential knowledge and skills required by the medical geneticist include the ability to:

a) establish a diagnosis;
b) interpret the role of the family history and assess the mode of inheritance;
c) know the indications for and how to interpret the results of genetic tests;
d) evaluate risk for the individuals and family relatives;
e) provide information on possible procreation choices;
f) discuss long term disease outcome.

26. Education and training should be provided in order to allow the medical geneticist to be able to communicate genetic information to enable patients to make informed decisions about themselves and where appropriate to raise awareness about possible implications for family members.

27. Education and training should be delivered in order to allow the primary care provider (general practitioner) to have knowledge of the most common chromosome abnormalities and monogenic disorders that are prevalent in the population concerned. This should include risk assessment, patient communication, a basic knowledge of pharmacogenomic developments, and the capacity to interface with other levels of care relevant for genetics, i.e. genetic labs, tertiary care. The general practitioner should also be aware of the indications and availability of predictive and prenatal diagnostics in order to adequately refer patients and their relatives to more specialised services when appropriate. Courses on basic medical genetics should be accessible to those in primary care physician training.

28. Institutions with medical geneticists and with a sufficient density of cases should offer education programmes in medical genetics. Curricula should be harmonised within Europe and preferably be of 5 years duration. Specialists in medical genetics must be competent in genetic diagnosis but must also be proficient in genetic counselling, and in the management and care of patients and their families.

29. The genetic laboratory scientist is responsible for genetic testing and for producing results of laboratory studies for the clinician to examine. The genetic laboratory scientist should be aware of the significance and the implications of the results of genetic tests. His/her curriculum should include basic medical genetics in parallel with specific technological education.

30. The curriculum of the genetic counsellor should include basic genetics, interpretation of pedigrees, risk assessment and communication skills.

31. The curriculum of specialists in other medical fields, for example internal medicine, paediatrics, neurology and ophthalmology, should ensure familiarity with clinical diagnosis and management of genetic disorders relevant for the respective speciality. During the specialist's training there should be an opportunity to spend some time, e.g. six months, under the supervision of a medical geneticist in order to be acquainted with principles of the genetic disorders related to the medical speciality.

32. Rapid progress made in the field of medical genetics has led to an increased demand for genetic services. There is a lack of trained medical geneticists and health care professionals to deliver the education and training. Governments should ensure that educational and research institutions are adequately equipped to meet the growing demand for health care professionals trained in medical genetics.

33. Specialist nurses may play an important role if trained in medical genetics. Training should be available to nurses who are able to provide basic genetic counselling and support management for the affected individuals and their families.

34. Practising clinical geneticists, genetic scientists and other health care professionals should receive continuing education in medical genetics. Continuing education should also equip primary care clinicians to provide genetic services and to enable these clinicians to recognise when referral to a medical geneticist is warranted. There is a need to secure a sufficient number of teaching personnel to enable medical geneticists and health care professionals to keep abreast with this rapidly expanding field.

III. Research in the field of medical genetics and genetic services

35. Genetic services encompass a broad range of activities including clinical diagnosis of genetic disease and syndromes, analysis of complex disorders, molecular and cytogenetic testing, management and treatment of genetic disorders. As in many other fields of medicine, progress depends on developments and outcome of research. Governments should encourage research in the field of medical genetics.

36. There are many areas of medical genetic research which are likely to improve the existing genetic services. These include:

a) research in natural history of rare monogenic disorders, especially genetic syndromes, which may require researchers to network closely with clinicians across Europe;

b) research on models of genetic counselling, care and management of patients and their families;

c) study of the psychological and social consequences for patients and their families with genetic disease;

d) improvement of molecular and cytogenetic diagnostic tests in order to improve clinical diagnosis and provide more accurate diagnosis and genetic counselling for the patient and their relatives;

e) establishment of bio-banks both for specific genetic disorders and population-based studies on molecular susceptibility profiles of common diseases;

f) ethical, legal and social issues raised by advances and developments in genetic medicine;

g) research in public health genomics where the discipline of public health is brought together with the knowledge from genetic and molecular science for the benefit of the health of the population.

37. There are other areas of research to be conducted in order to improve the quality of genetic services:

a) application of genomics to increase the knowledge of the causation of disease and to unravel the genetic susceptibility profiles of multifactorial disorders;

b) gene therapy is an important area of research, especially in monogenic disorders. It offers the potential to cure or alleviate monogenic disorders by introducing new genes into the body (somatic cells) to replace faulty genes. For example, severe combined immune deficiency (SCID), a life-limiting disease, with death in the first few years of life, has been treated successfully by replacing the defective gene. Currently, clinical trials are ongoing for several other monogenic disorders, such as Duchenne muscular dystrophy, haemophilia and Leber's amaurosis congenita;

c) research in pharmacogenomics could make the use of medicines more effective. The development of molecular profiles of individuals will allow the identification of patients who will respond successfully to the treatment, of those who will react in an adverse manner and of those who will not respond to a particular drug. This should lead to 'personalised treatment'. Successful research in this area would be invaluable in identifying medicines which are not beneficial because of a related genetic toxicity;

d) genomic research in understanding the pathogenesis of multifactorial disease will be important for public health, especially in developing public health policies and services for benefitting the health of the population. A main area will be the development of disease prevention programmes directed to susceptible individuals and families and indeed to sub-population groups with a genomic risk profile;

e) research in the field of epigenetics will unravel many of the unexplained phenomena in genetics today and are therefore of great importance.

38. Genetic registers for specific genetic disorders are important for research purposes and the development of new treatments.

39. Member states should encourage collaboration and networking of research in medical genetics across Europe and internationally. This is especially true for rare genetic disorders so that a significant body of knowledge and experience is accumulated and introduced into the genetic service as rapidly as possible. It also applies to research development of bio-banks where researchers need to gain access to biological materials and relevant data, taking into account data protection concerns in conformity with the provisions of Recommendation (2006)4 on research on biological materials of human origin.

IV. Medical investigations in genetics

Genetic laboratory tests

40. A genetic laboratory test is to be understood within the meaning of genetic tests as defined in Article 2 of the Additional Protocol which reads as follows:

> ***Article 2 – Scope***
> *1 This Protocol applies to tests, which are carried out for health purposes, involving analysis of biological samples of human origin and aiming specifically to identify the genetic characteristics of a person which are inherited or acquired during early prenatal development (hereinafter referred to as "genetic tests").*
> *2 This Protocol does not apply:*
> *a to genetic tests carried out on the human embryo or foetus;*
> *b to genetic tests carried out for research purposes.*
> *3 For the purposes of paragraph 1:*
> *a "analysis" refers to:*
> *i chromosomal analysis,*
> *ii DNA or RNA analysis,*
> *iii analysis of any other element enabling information to be obtained which is equivalent to that obtained with the methods referred to in sub-paragraphs a.i. and a.ii.;*
> *b "biological samples" refers to:*
> *i biological materials removed for the purpose of the test concerned,*
> *iii biological materials previously removed for another purpose.*

The requirement that the test involves the analysis of a biological sample excludes as such the collection of genetic information through family history.

The notion of “genetic test” is based here on two elements: the method used and the purpose of the test. It is to be understood as a procedure including removal of biological material of human origin, where relevant, as well as the analysis of the personal information obtained there from. This procedure aims specifically to identify genetic characteristics of a person which are inherited or acquired during early prenatal development. These genetic characteristics cover those already present in the gametes of the parents and therefore transmitted by the latter, as well as those which appear during the early stage of prenatal development before the differentiation of the germ line. It is sometimes referred to the genetic characteristics inherited or acquired during early prenatal development as “genetic characteristics transmissible to descendants”. The genetic modifications acquired during lifetime by only certain somatic cells due for example to external factors in the environment, are therefore not covered.

41. The use of genetic testing in health care is growing at a staggering rate. It is widely used in diagnosis of monogenic disorders and may be indicated for other types of disorders. At present, genetic testing in multifactorial diseases and in pharmacogenomics is small but will undoubtedly grow in the next 5-10 year. Genetic testing for familial cancers such as breast and bowel cancers has markedly increased. Future discoveries of genes related to familial cancer will undoubtedly lead to an increase in the volume of tests. As it becomes possible to identify specific molecular patterns related to common diseases, these tests will put larger demands on genetic services because of higher patient numbers.

42. As the public’s awareness increases and the pace of new discoveries mounts, there will be a demand for incorporating new developments into genetic services and medicine in general. These developments will include:

a) increasing incorporation of genetic clinics in primary care and in familial cancer clinics;

b) improving molecular cytogenetic techniques that add precision to the diagnosis (dosage analysis for gene duplications and deletions);

c) incorporating more monogenic disorders into screening programmes;

d) introducing and extending rapid prenatal diagnosis;

e) organising the genetic management of complex diseases in collaboration with other medical disciplines;

f) using pharmacogenetics in health care in collaboration with other medical disciplines and the pharmaceutical industry;

g) public health genomics.

43. Several types of genetic tests are available for different clinical purposes:

a) diagnostic genetic tests: undertaken on a symptomatic individual to establish, confirm, refine or exclude a clinical diagnosis including tests to identify the causative mutation/chromosomal change so that genetic testing in other situations may be carried out (e.g. prenatal diagnosis or testing of relatives);

b) predictive genetic tests: undertaken on asymptomatic individuals to:

– identify a mutation which will lead to future disease - tests predictive of a monogenic disease;

– identify a mutation which may lead to future disease - tests serving to detect a genetic predisposition or genetic susceptibility to a disease;

– identify the subject as a healthy carrier of a gene responsible for a disease: a carrier is an individual who is heterozygous for a gene mutation for a disorder inherited in an autosomal recessive or X-linked manner or a balanced chromosomal alteration. Carriers rarely have symptoms related to the defective gene or chromosomal aberration but there may be implications for the health of their children;

c) prenatal genetic tests are performed to diagnose or exclude the presence of a mutation in a foetus during pregnancy;

d) preimplantation genetic testing are tests performed to diagnose or exclude a genetic condition in an embryo obtained through *in vitro* fertilisation;

e) genetic screening programme tests: screening tests are carried out on a population or a defined sub-group of the population for systematic early detection or exclusion of a genetic disorder, the genetic predisposition or resistance to a disease, or to determine carriers of a gene variant which may produce disease in children. The logic underlying the decision indication to screen is population-based and not individual-based (see Chapter V, below).

44. Genetic tests are one category of medical tests and are not fundamentally different from other laboratory tests in many aspects; however genetic tests may also have implications for other family members. Genetic laboratory tests should be subject to the general principles governing other medical tests. As genetic tests include many difficult ethical aspects special considerations are necessary. Information and genetic counselling should be provided in conformity with the provisions of Articles 8 and 9 of the Additional Protocol. In Europe, the European Society of Human Genetics (ESHG) has produced guidelines for professionals (www.eshg.org) and the Network for Excellence EuroGentest (www.eurogentest.org) is working to develop the necessary infrastructure, tools, resources, guidelines and procedures leading to the establishment of harmonised quality genetic testing services in Europe.

45. There are clearly important components of genetic testing best practices:

a) clinical validity of genetic testing refers to the measure of accuracy, which includes clinical sensitivity, specificity and predictive value, with which the test identifies or predicts a clinical disorder;

b) clinical utility is the ability of the test to provide information that is of value in a clinical situation;

c) the method of the test often depends on the particular laboratory undertaking the investigation. The required result may be obtained by different methods and there may be no absolute best method;

d) the purpose of the test could be for a variety of reasons, such as diagnosis in a symptomatic person, predictive testing, carrier detection, prenatal diagnosis, and preimplantation genetic testing;

e) the quality of the test depends on many factors, such as the quality system of the laboratory, quality control programmes and accreditation;

f) information about the test provided by the laboratory scientist to the clinician can affect the use and interpretation of the results. In ‘over the counter’ tests, the information is provided directly to the client;

g) genetic counselling is important in genetic testing.

46. There is a need to structure, harmonise, and improve the overall quality of genetic testing across Europe. It is essential to develop and maintain the web-based database(s) to assist clinicians and scientists to find reliable high quality tests for their patients and families. Governments should be aware of the importance of such database(s) and consider providing resources for their maintenance.

47. The communication of the availability and of results of genetic testing requires careful consideration. The public needs to have information about testing to prevent raising false expectations and to provide a realistic view of genetic testing. This can be achieved through education in schools and through the media. As research produces genetic tests for common multifactorial disorders and particularly with the production of tests supplied direct to the public (“over the counter” tests), informing the public should be an ongoing process.

48. Equitable access to genetic counselling and genetic tests should be the norm and should be independent of geography or financial means.

49. Genetic tests should be performed by high quality service laboratories. However some genetic tests may be performed by research groups, in collaboration with service laboratories.

50. Genetic testing for rare genetic diseases is not universally available throughout Europe. However, samples of extracted DNA for mutation or marker identification can be sent by post to specialist or research laboratories specialising in particular rare diseases. The referring clinician can identify the laboratory undertaking the test for these rare diseases from, for example, Orphanet (www.orphanet.org).

51. Genetic variants of DNA are common in various populations and a DNA profile of the variant patterns may be associated with common multifactorial disorders or may also predict a patient's response to medication. It is hoped that specific patterns will be identified which will enable the clinician to provide the correct medication for the patient (personalised medicine) and avoid serious side effects of the particular drug (pharmacogenetics).

Other examinations for genetic disorders

52. The medical geneticist employs a wide range of investigations to establish a clinical diagnosis in patients. These investigations include physical and phenotypic examination, pedigree analysis, conventional laboratory tests, imaging techniques and electro-physiological measurements. With the development of genetic tests (see above) these are used to confirm the diagnosis in order to provide accurate genetic counselling.

53. The medical geneticists' team should have adequate capabilities including the necessary IT skills to interrogate databases on the internet such as OMIM (Online Mendelian Inheritance in Man, http://www.ncbi.nlm.nih.gov/omim/).

54. The OECD survey on quality assurance and proficiency testing (2005) compared practices in individual countries in order to inform international action in setting standards and developing guidelines for practice. Based on the survey results, the report puts forward recommendations for action for better quality assurance and proficiency of molecular genetic testing. It shows, for example, that requirements for licensing and accreditation/certification of diagnostic molecular genetic testing laboratories have not been sufficiently followed in OECD member states in a systematic way. Considerable variations exist in mechanisms of licensing, certification and accreditation, including the standards by which tests are performed, results are reported, and the qualifications for laboratory personnel.

55. There are two types of examination: one which is very broad, even including a family history analysis. For this examination stringent control would perhaps not be appropriate. A second type of examination would imply laboratory tests, where certification is desirable. Regular systematic surveillance would be required, and, where appropriate, certification or accreditation. In practice, the health facility should be subject to regular systematic surveillance, and where appropriate, be certified or accredited by a competent authority.

V. Organisation of genetic screening programmes

56. Genetic screening must be distinguished from individual genetic testing. Screening refers to public health programmes aimed at either whole populations of asymptomatic individuals or at sub-populations in which there are individuals at an increased risk of a genetic disorder. It is performed for the systematic early detection or exclusion of a hereditary disease, the predisposition to such a disease or to determine carriers of a predisposition that may produce a hereditary disease in offspring. For example, screening the whole neonatal population for phenylketonuria or screening a sub-population such as Ashkenazi Jews for carriership of Tay-Sachs disease.

57. Genetic screening programmes should only be implemented after fulfilling well-defined criteria as specified in Article 19 of the Additional Protocol. Screening programmes are only justified for genetic disorders of significant severity, frequency and health impact. The evaluation of the programme should consider issues such as frequency of the disease in the community, scientific validity of the test procedure, effectiveness, health relevance to the population or sub-population, and the availability of preventive measures or treatment in respect to the disease which is subject to screening.

58. Screening programmes have limitations. While screening programmes have the potential to save lives or improve the quality of life through early diagnosis of serious genetic disorders, they can cause anxiety and distress in the participants. Some individuals could be wrongly reassured by a false negative result, while on the other hand a false positive result may lead to unnecessary worry and follow-up of the patient. In addition, as many screening programmes are for genetic diseases which may be inherited, there are implications for other family members (blood relatives). For example, in a neonatal screening programme the identification of a baby with phenylketonuria would mean that the parents have an increased risk of another affected infant in future pregnancies. Genetic counselling should be available to all families in which the screening programme identifies an affected individual.

59. The screening programme must take into account the issues of privacy of the personal information about the health status of the individual, and release of personal information about the patient must not be revealed without the patient's written consent. Safeguards must be in place to prevent stigmatisation and discrimination of the individual. The same protection requirements should apply to screening programmes as to diagnostic genetic tests.

60. Criteria for introducing genetic screening programmes should include ethical considerations as well as public health ones. Genetic screening goals and the target population must be well defined; stringent laboratory quality control, with limits of results clearly delineated; the confidentiality of the information protected by authorities; procedures to protect individual and family privacy established in advance; participation voluntary; genetic counselling offered and educational programmes in place; and long-term outcomes monitored and evaluated.

VI. Health technology assessment

61. New health-related technologies are evaluated in order to assess their clinical benefit in terms of health outcomes, and to consider the effect on the care of individuals being tested. This evaluation includes safety and efficacy, and its depth depends on the nature of the technology and regulatory requirement. The main outcome of any genetic test, as in any diagnostic test, is information. Any subsequent health outcome gain will depend on the decision(s) made at the individual level and by the effectiveness of further care.

62. There is a clear need to obtain reliable information on the effectiveness of genetic testing services for different diseases, compared to other treatments or prevention strategies. Assessing the economic impacts of genetic tests should consider the health care system as a whole. This implies the assessment not only of the clinical effectiveness of genetic tests, but also the impact on the health care system in terms of delivery of services, human resources or possible side-effects on patients' and relatives' health.

63. Technology assessment is a well-developed field in many European countries. A Technology Assessment process might be described as a scientific, interactive and communicative process which aims to contribute to forming public and political opinion on economic and societal aspects of science and technology. It encompasses different levels of analysis. At its first level, the safety, efficacy and effectiveness of technology are addressed to establish its clinical utility. A second level of assessment is comprised of an economic appraisal. This is usually applied when decisions are under consideration regarding the acquisition of new technology, health insurance coverage or the scope of a health benefits list. There is also a third level: to address ethical, legal and social issues (ELSI). Genetic testing raises new methodological difficulties at all three levels, but above all when considering the full range of costs, benefits and risks and the ELSI dimension. ELSI may be the primary concern in some cases, for instance, when dealing with prenatal tests or screening.

64. Additionally it should be remembered that genetic services may be subject to quality assurance, accreditation and suchlike as appropriate, as any other clinical facility or service.

65. The rapid pace of scientific discoveries and developments, and thus the availability of tests with the resulting implications for healthcare delivery and for resource requirement considerations should be borne in mind. Public policy facilitating institutional cooperation at national and international levels, including active scientific information sharing, general networking of clinicians, scientists and their institutions, mobility of professionals, and easy access to special tests or services should be considered. There is a technology assessment approach named “horizon scanning” which aims to detect technologies and innovations that are close to entering clinical use: it has proved to be useful in diverse contexts of health policy. Access to this assessment approach is most desirable for both policy-making and for health planning.

66. The actual direct cost of acquiring or providing a genetic test could be comparatively small with regard to the aggregate costs of prevention and treatment for the target population. Policy makers should evaluate all economic and ELSI aspects of genetic services at least for both resource allocation and benefits/coverage considerations. Furthermore the need for extra educational efforts should be considered, both for professionals and for society at large to guarantee a minimum of quality and rigour in the process of allocation/coverage considerations.

VII. Public awareness

67. The pace of genetic research is accelerating and advances made in this field feed into the practical application at the level of clinical medicine. This opens up ever increasing opportunities to bring about improvements in health care. The public may view these developments positively but also with a degree of anxiety and apprehension.

68. Members of the civil society should be involved as partners in the development of open and transparent policies on genetics. Several means could be used, namely with surveys to identify attitudes of the public towards current and new developments, and with public focus groups and citizens’ juries.

69. Programmes can be offered to increase public awareness and understanding of human genetics. Such programmes should be introduced during health education in schools. They would need to be flexible to meet the fast moving field of medical genetics.

70. The printed and broadcast media, as well as electronic-based media and internet, play an important part in the education of the public. Although accessing information may be direct, data portrayed may also be inaccurate, incomplete or biased, which may create confusion. In order to guard against this, Governments should take appropriate measures to facilitate access for the public to objective general information on genetic tests, including their nature and the potential implications of their results. Research institutions should be encouraged to develop websites especially devoted to informing the public of the nature of research developments and also the ethical and social implications of their research.

71. Consideration should be given to the development of common platforms of genetic knowledge where collaboration takes place between genetic services, research institutions, patient support groups, and are open to the public. Such platforms will bring together a strong combination of expertise from many disciplines. Not only will such developments facilitate the translation of advances in genetics into the clinical environment but they will also provide opportunities for collaborative medical genetic research. Because of the involvement of the public and patient support groups, there will be a ready opportunity to ascertain the public’s attitudes to new and novel developments. Common platforms of genetic knowledge might also facilitate the involvement of the private sector.